Rhabdoid tumor predisposition syndrome

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Published in Atlas Database: March 1999

Online updated version : http://AtlasGeneticsOncology.org/Kprones/rhabdKpronID10051.html

DOI: 10.4267/2042/37522

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Clinics

Phenotype and clinics

Scarcely known, with only three reports:
- two siblings with a paravertebral malignant rhabdoid tumor in the first year of life and a poor outcome; no family history,
- a patient with renal rhabdoid tumors associated with tumors of the central nervous system,
- four children with germ-line mutations of INI1, three with renal rhabdoid tumors and one with an atypical teratoid tumor of the brain (out of 18 atypical teratoid and rhabdoid tumors studied).

Genes involved and proteins

\( hSNF5/INI1 \)

Location

22q11.2

Mutations

Germinal: are found in this syndrome.

Somatic: mutation and allele loss events in sporadic rhabdoid tumors are in accordance with the two-hit model for neoplasia, as is found in retinoblastoma.

References


Fort DW, Tonk VS, Tomlinson GE, Timmons CF, Schneider NR. Rhabdoid tumor of the kidney with primitive neuroectodermal tumor of the central nervous system: associated tumors with different histologic, cytogenetic, and molecular findings. Genes Chromosomes Cancer 1994;11:146-152


This article should be referenced as such: